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*Brief Clinical Report*

# Linear Disruption of Umbilical Cord: A Rare Anomaly of the Cord Associated With Acute Fetal Distress and Perinatal Death/Profound Psychomotor Retardation

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**We report on a non-malformed child with severe microcephaly and profound psychomotor delay. Review of the delivery/birth records documented descriptions consistent with linear disruption of the umbilical cord. This rare anomaly typically leads to acute fetal distress and perinatal death. Severe microcephaly and psychomotor delay without other anomalies should prompt a careful review of the delivery/birth records to search for umbilical cord descriptions consistent with this diagnosis.** 

**KEY WORDS: umbilical cord, Wharton jelly, linear disruption, microcephaly, psychomotor delay**

## INTRODUCTION

Linear disruption of the umbilical cord is a rare anomaly with a prevalence of less than 1 in 3,500 during a 5-year period at one hospital [Labarrere et al., 1985]. It is characterized by absence of Wharton jelly, mainly around the arteries rather than the vein and absence of the membrane (Fig. 1). The "naked arteries may appear like garlands" [Blackburn and Cooley, 1993] and have a dark coloration. The affected area of the cord appears thinner. The vessels may be exposed throughout the whole cord or segmentally.

Linear disruption of the umbilical cord is associated with acute onset of fetal distress and usually perinatal death [Labarrere et al., 1985; Clausen, 1989]. Compression of the "naked" cord arteries has been thought to account for the clinical symptoms [Labarrere et al., 1985].

## CLINICAL REPORT

A 3$^{9}/_{12}$-year-old girl with severe microcephaly and profound psychomotor retardation was evaluated due to parental concern regarding recurrence risk. History indicated acute cessation of fetal movements 1 week before the expected date of confinement. A female infant of size appropriate for gestational age was delivered by emergency C-section. Birth weight was 3,450 g, length was 48.5 cm, and OFC was 35.0 cm. Delivery notes indicate the umbilical cord had "scores of twists," was almost totally devoid of Wharton jelly, and was entangled around the baby's body and between the legs and perineum. Part of her umbilical cord appeared darker and thinner. According to the pathology report, the umbilical cord had a maximum diameter of 0.8 cm (normal range at term is 1.0–1.2 cm). The full length of the cord was not received by the Department of Pathology. Cord insertion was "slightly eccentric." Three well formed vessels were present. The placenta was discoid in form, measured 18 × 15 × 1.5 cm in greatest dimensions, and weighed 515 g. The fetal surface of the placenta showed a submembranous irregularly shaped grayish white area measuring 4 × 3 cm in the greatest dimension. On section, the area was confined only to the membrane. On microscopic examination, there was an occasional small focus of hemorrhage and several small foci of infarction. The infant was severely distressed at birth, but she responded to resuscitation efforts (Apgar scores $0^{1}$, $0^{5}$, and $1^{10}$). A brain CT scan done at age 2 weeks showed the effects of hypoxic brain insult. A repeat brain CT scan at 2½ months of age showed diffuse supratentorial brain atrophy indicating leukomalacia likely secondary to anoxic brain insult and ventriculomegaly with microcephaly. An EEG at age 3½ years was consistent with seizures. Family history was noncontributory.

At 3$^{9}/_{12}$ years she had severe growth failure and microcephaly (OFC 42.0 cm). No minor anomalies were present. Skin was normal. Neurologic examination documented spasticity, psychomotor retardation, poor

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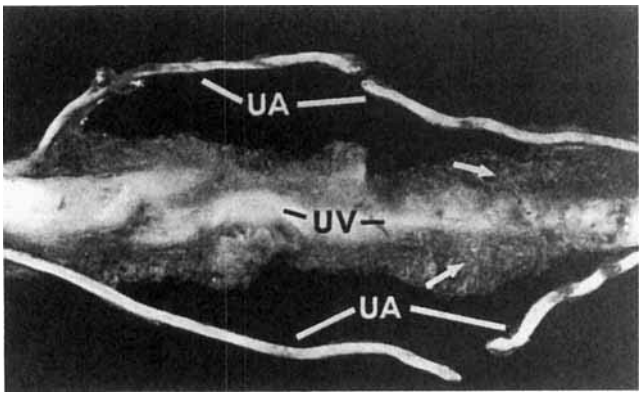


Fig. 1. Linear disruption of the umbilical cord showing "naked" umbilical arteries (UA) without supporting Wharton jelly. The umbilical vein (UV) continues to be surrounded by considerable Wharton jelly and matrix material (arrows). [From Blackburn and Cooley, Jr. (1993): The umbilical cord. In Stevenson RE, Hall JG, Goodman RM (eds): "Human Malformations and Related Anomalies," Vol II. New York: Oxford University Press, p 1093.]

head control, inability to sit, and able to produce only open vowel sounds. Chromosomes and plasma amino acid composition were normal.

## DISCUSSION

Other than the brief description by Blackburn and Cooley [1993], linear disruption of the umbilical cord is not recorded in the clinical genetic literature. Based on the finding of an unusually thin umbilical cord devoid of Wharton jelly and a photograph taken by the parents, the diagnosis of linear disruption of the umbilical cord was made. Since most cases with umbilical cord anomaly are sporadic events, and a familial linear umbilical cord disruption to our best knowledge has not been reported, the recurrence risk is likely to be very small (<1%).

The cause of the anomaly is unknown. It has been postulated that linear disruption of the umbilical cord is 1) a more severe form of umbilical cord cyst formation [Bergman et al., 1961], 2) an incomplete fusion of the amniotic covering and the mesenchyme of the umbilical cord during early development, or 3) a hypoplasia of the amniotic covering with a secondary loss of Wharton jelly [Labarrere et al., 1985]. Finally, since meconium stained amniotic fluid is a frequent finding, the loss of Wharton jelly could also be the result of a proteolytic digestion of the amniotic membrane around the cord by meconium enzymes [Blackburn and Cooley, 1993]. Our case does not support the latter theory. Meconium was not noted in the delivery or neonatal records.

An anomaly of the umbilical cord, such as linear disruption, thrombosis [Clausen, 1989], coarctation [Tavares Fortuna and Lourdes Pratas, 1978], or torsion [Herman et al., 1991] can account for acute fetal distress, perinatal death or severe microcephaly and profound psychomotor delay. We suggest that review of delivery/birth records can be helpful in evaluation of cases with severe microcephaly. Knowledge about this anomaly may prevent unnecessary and expensive laboratory analyses searching for an etiology and may provide the parents with the reassurance of a small recurrence risk.

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